One Medicine One Pathology: 2nd annual CASIMIR Symposium on Human and Mouse Disease Informatics

UMLS and phenotype coding

Anita Burgun, Fleur Mougin, Olivier Bodenreider

INSERM U936, EA 3888- Faculté de Médecine, Univ. Rennes1
ISPED, Univ. Bordeaux, France
US National Library of Medicine, NIH, Bethesda, USA

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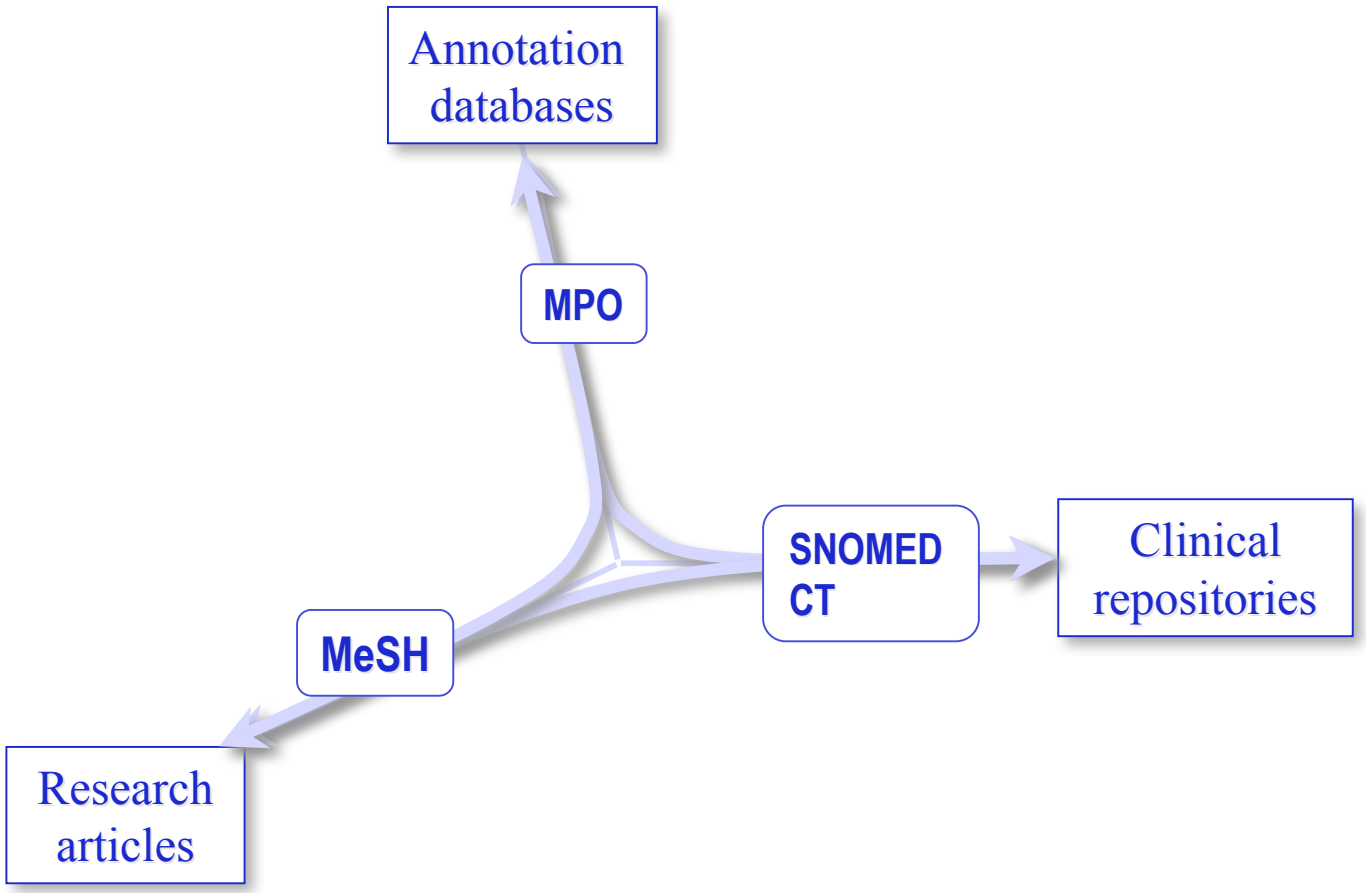
Phenotype coding



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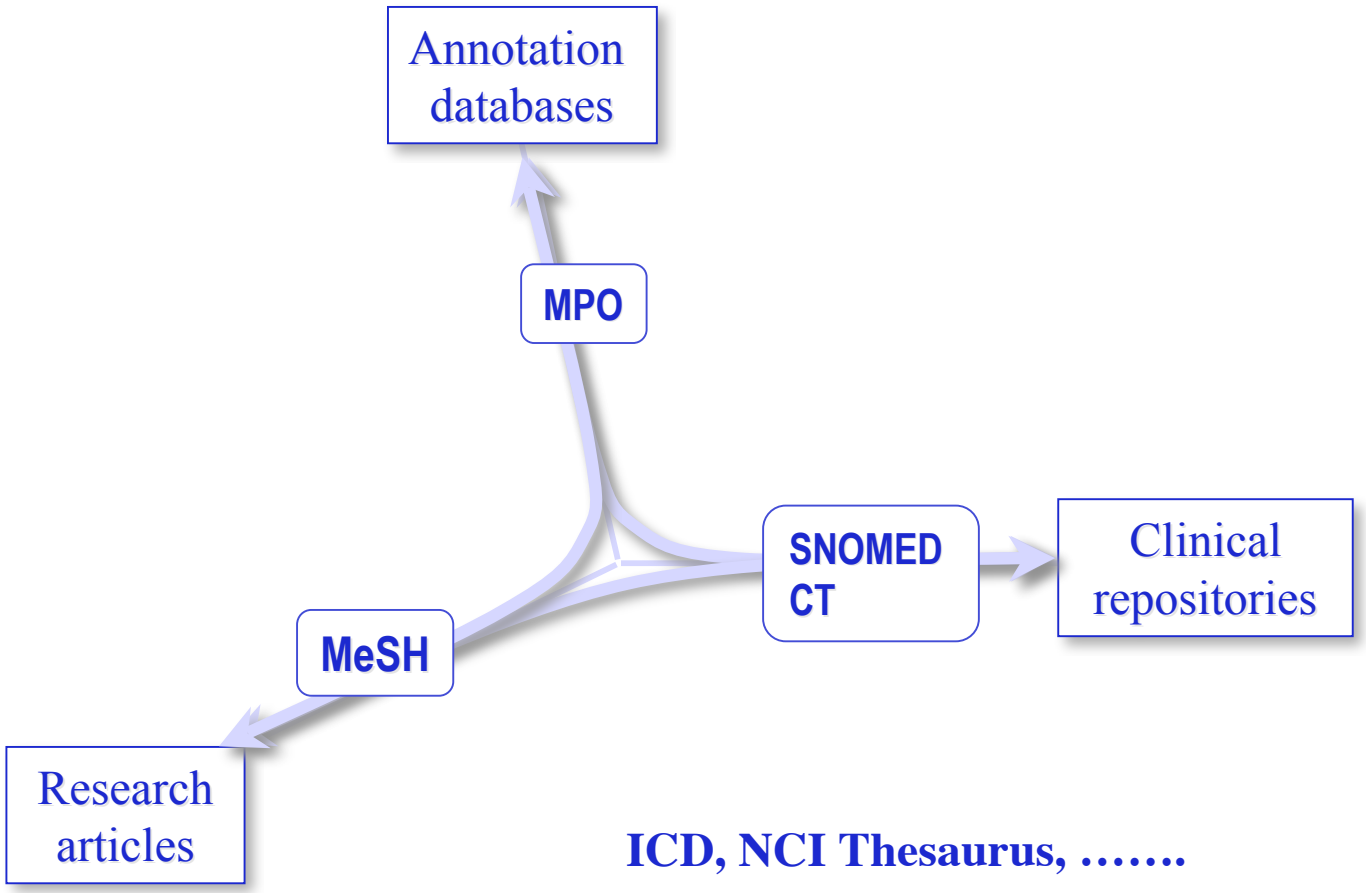
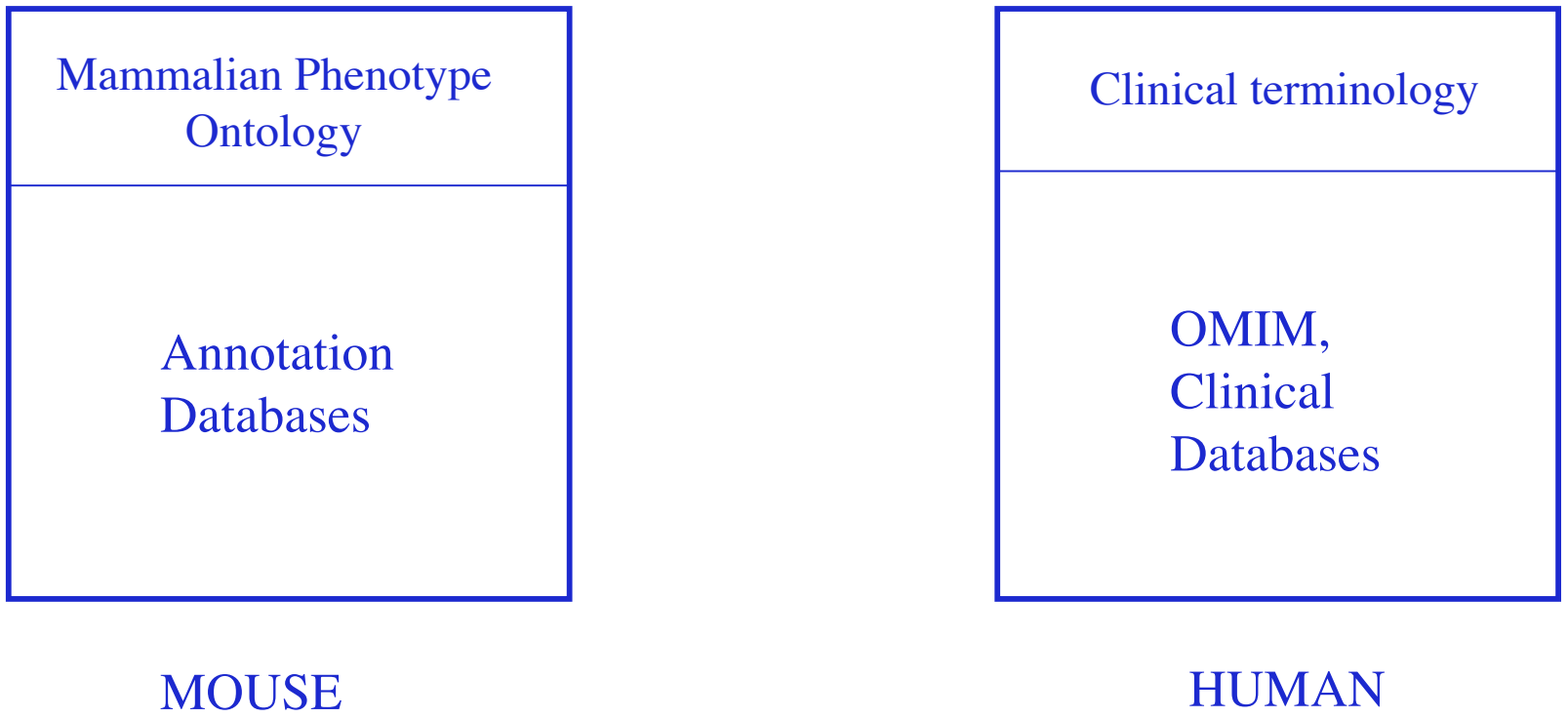


Phenotype coding: role of the UMLS



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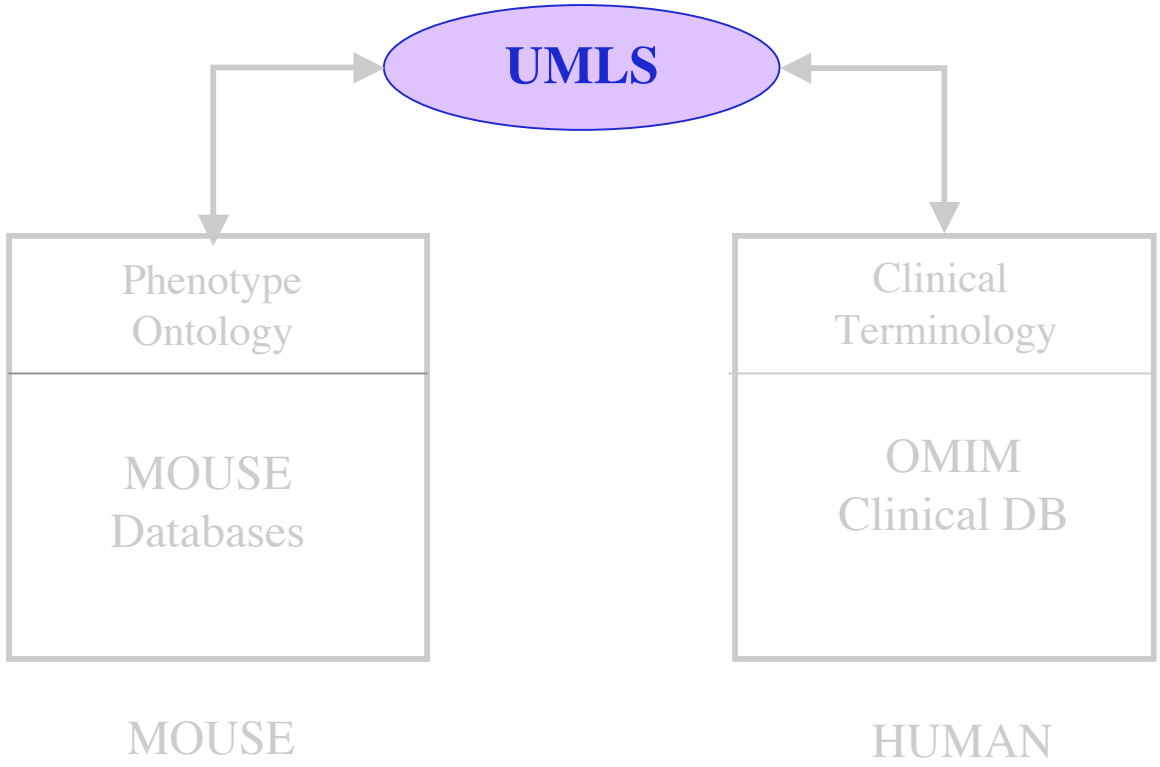

Unified Medical Language System

- Addresses heterogeneity issues
  - More than 100 source vocabularies
  - Unification
- Clusters terms into concepts
  - Metathesaurus: CUIs
- Organizes hierarchies
  - Metathesaurus: relations
- Categorizes concepts
  - Semantic Network: Semantic Types

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UMLS Metathesaurus

- Craniosynostosis in UMLS Release 2008AA
- Source vocabularies
  - ICD-10
  - ICPC
  - MedDRA
  - MeSH
  - OMIM
  - Read Codes
  - SNOMED CT
  - …..
- Definition (MeSH) : Premature closure of one or more sutures of the skull

- UMLS Metathesaurus
- CUI C0010278 Craniosynostosis (Preferred Term)
  - Craniostenosis (ICD, ICPC, OMIM, SNOMED CT)
  - Craniosynostosis syndrome (SNOMED CT)
  - Synostosis (cranial) (CRISP)
  - *Word phrases*
    - Premature closure of cranial sutures (MedDRA, SNCT)
    - Congenital ossification of cranial sutures
    - Congenital ossification of sutures
    - Congenital ossification of sutures of skull
    - Premature cranial suture closure (SNOMED)
  - *Abbreviations*
    - CRS, CSO, CRS1 (OMIM)
  - *More specific terms*
    - Craniosynostosis, type 1 (OMIM)

- Possible synonyms and related
  - Hurst syndrome (C0014077)
  - Christian syndrome 1 (C0795794)
  - SCARF (skeletal abnormalities, cutis laxa, craniostenosis, psychomotor retardation, facial abnormalities) syndrome (C0796146)…..

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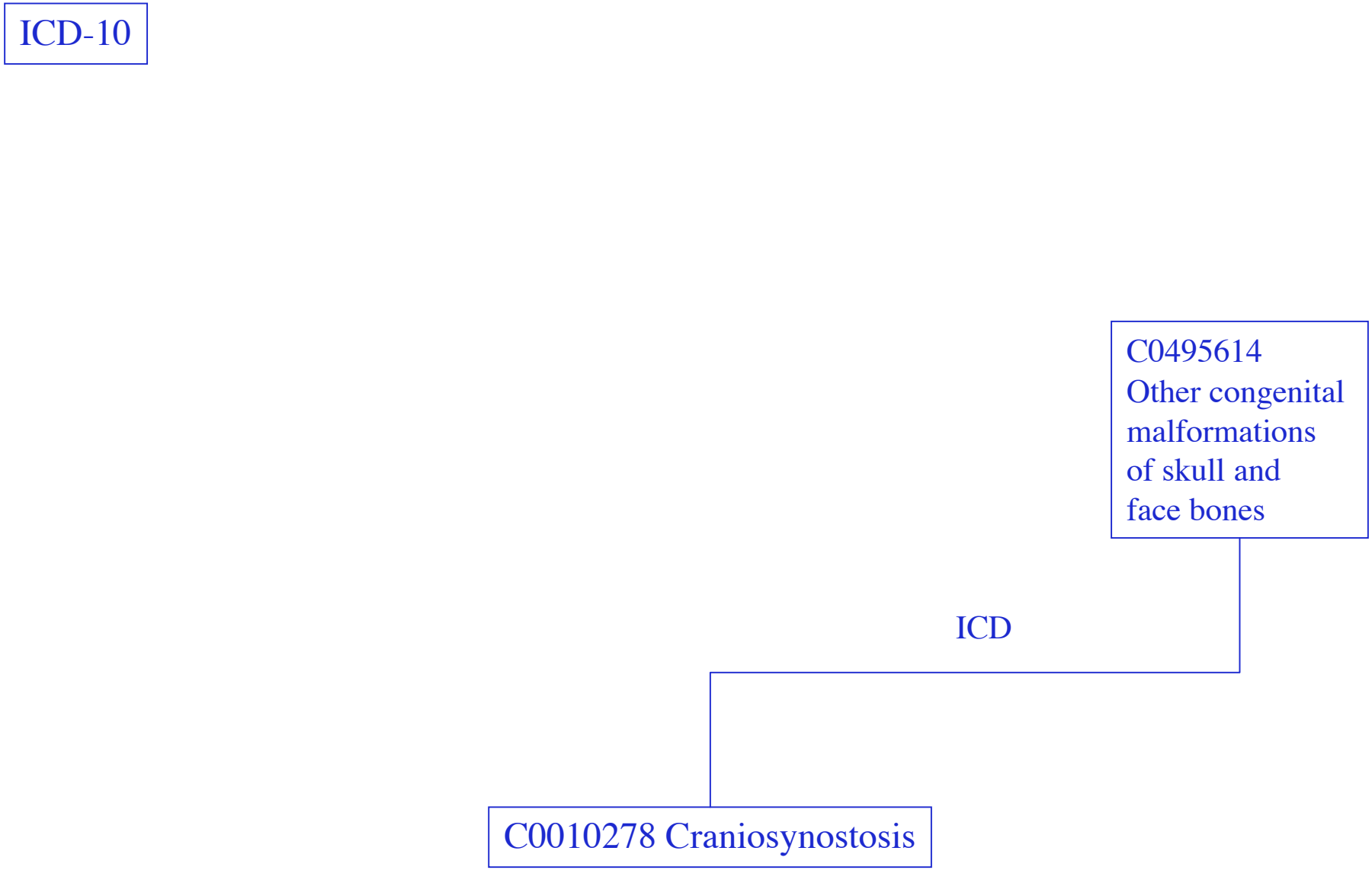
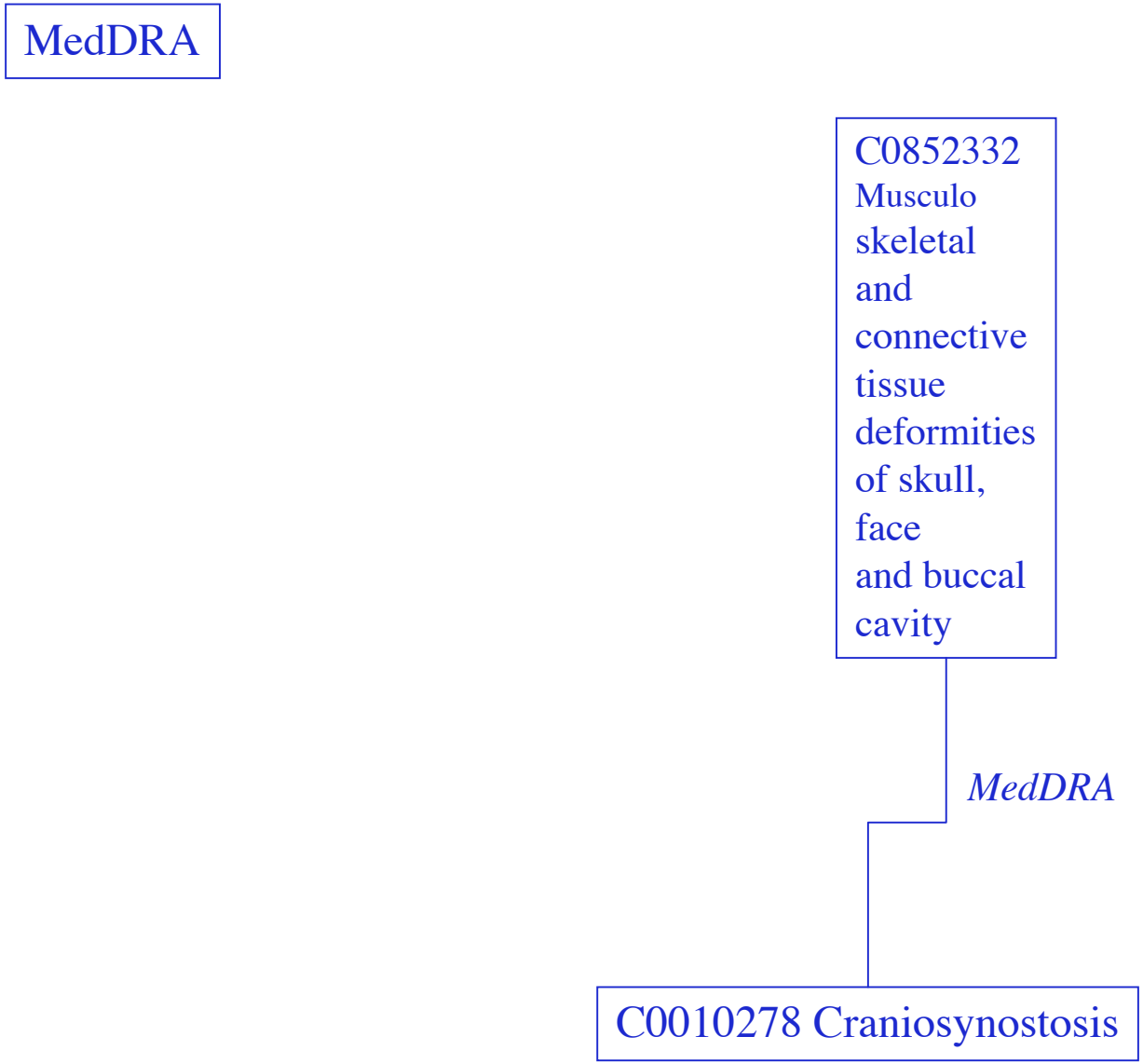
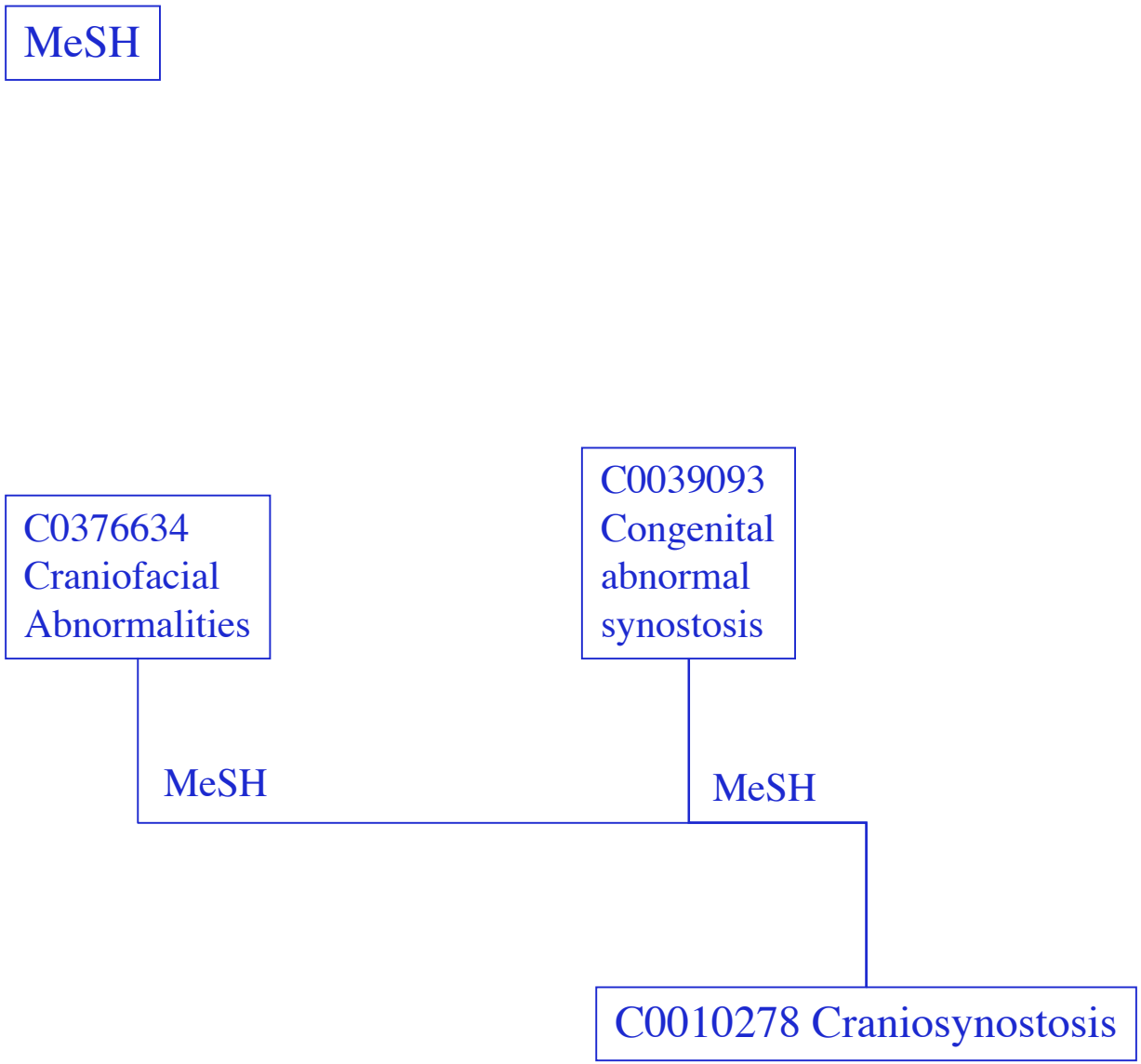
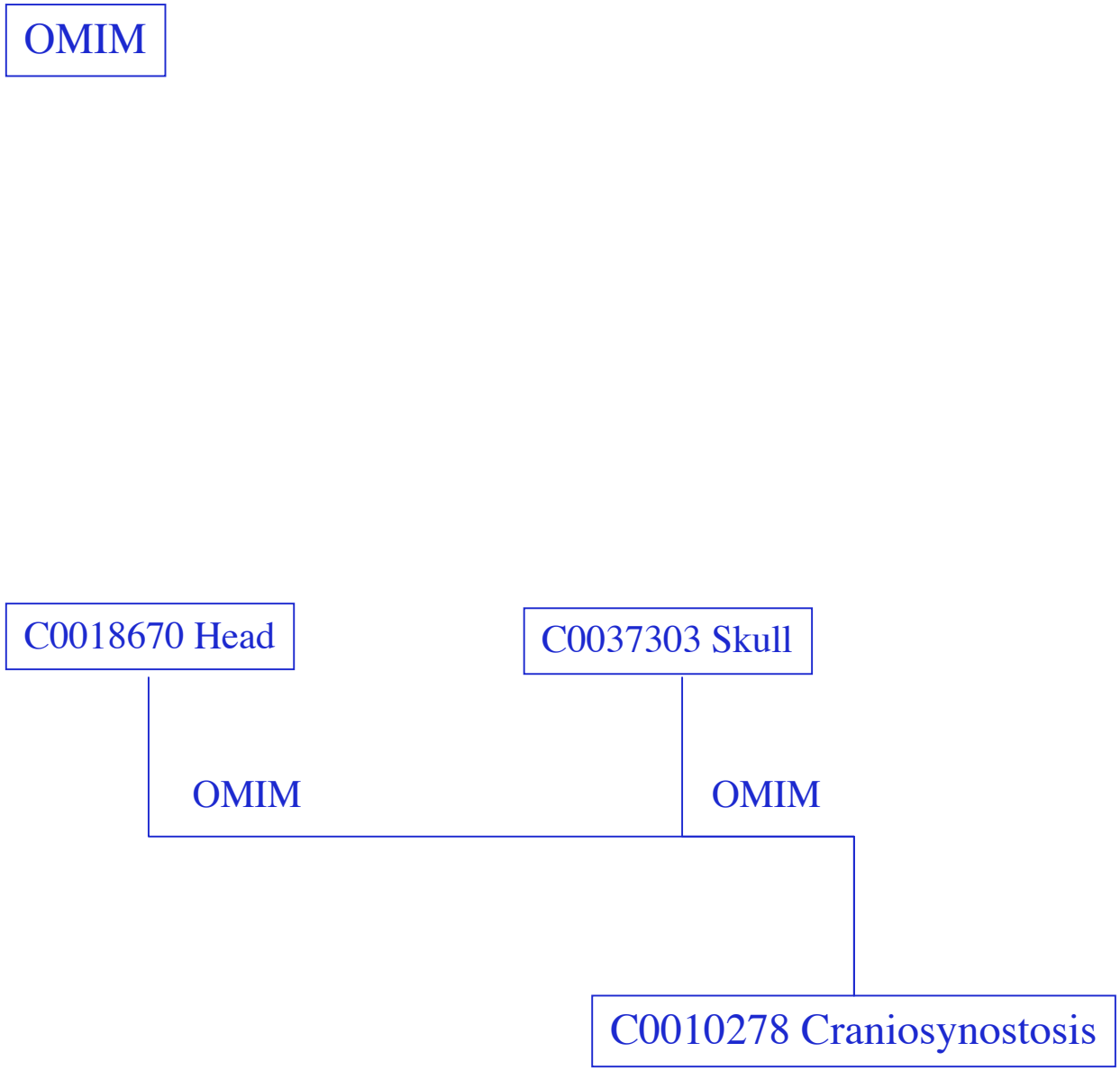
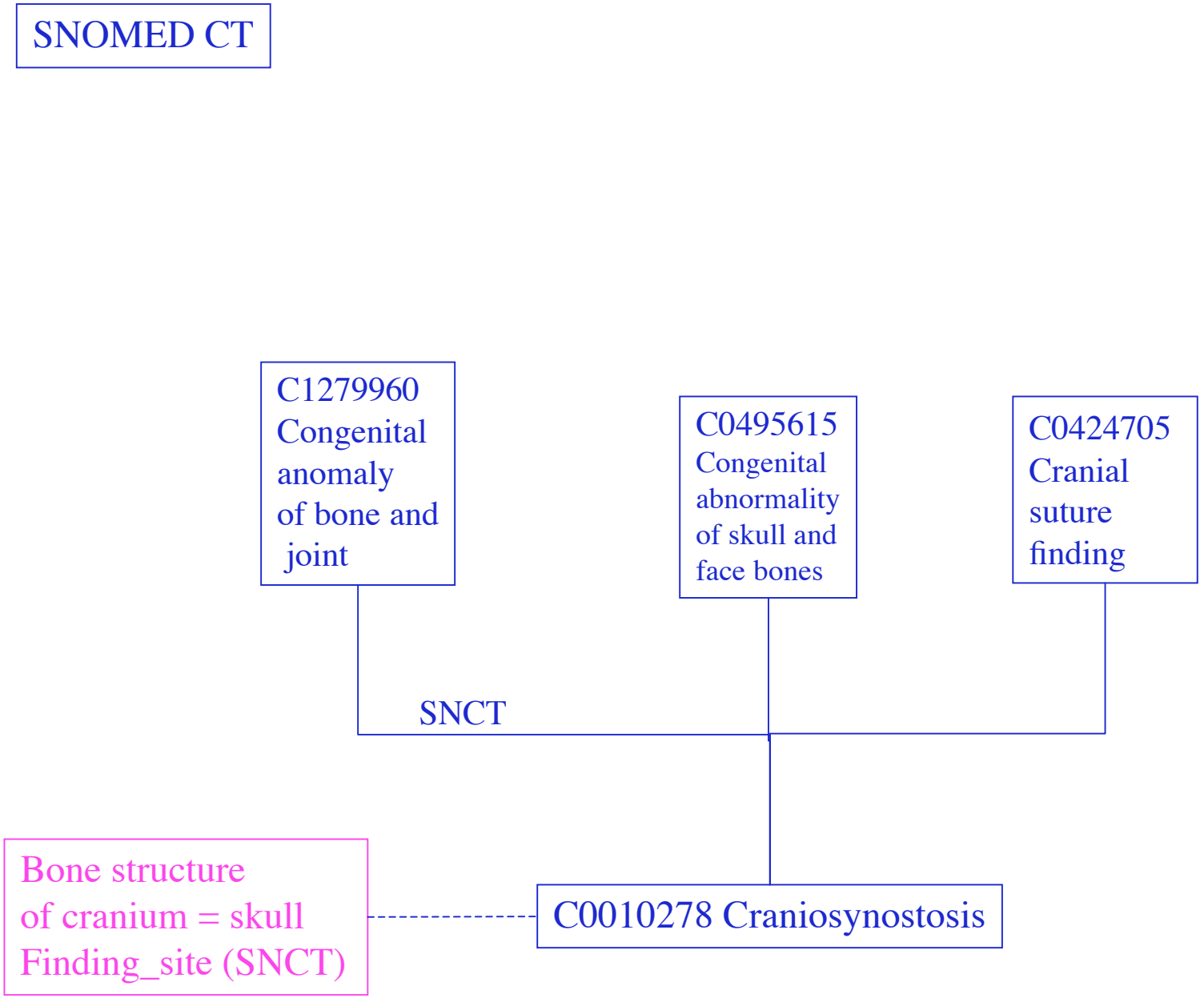
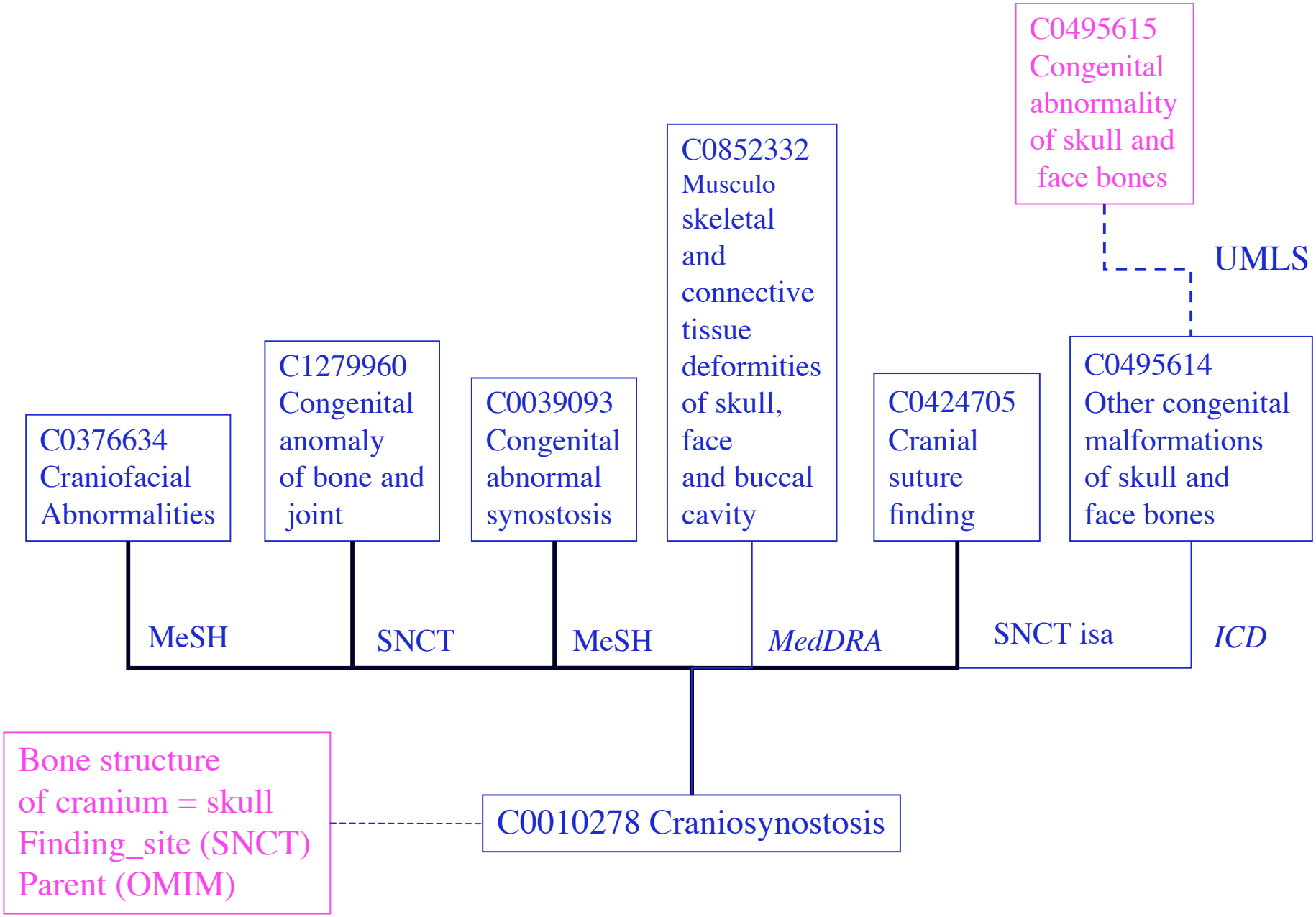

ICD-10
C0495614
Other congenital
malformations
of skull and
face bones
ICD
C0010278 Craniosynostosis


MedDRA
C0852332 Musculo skeletal and connective tissue deformities of skull, face and buccal cavity
MedDRA
C0010278 Craniosynostosis


MeSH
C0376634
Craniofacial
Abnormalities
C0039093
Congenital
abnormal
synostosis
MeSH
MeSH
C0010278 Craniosynostosis


OMIM
C0018670 Head
C0037303 Skull
OMIM
OMIM
C0010278 Craniosynostosis


SNOMED CT
C1279960
Congenital
anomaly
of bone and
joint
C0495615
Congenital
abnormality
of skull and
face bones
C0424705
Cranial
suture
finding
SNCT
Bone structure
of cranium = skull
Finding_site (SNCT)
C0010278 Craniosynostosis


C0495615
Congenital
abnormality
of skull and
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UMLS
C0852332
Musculo
skeletal
and
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tissue
deformities
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C0376634
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Other congenital
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Parent (OMIM)
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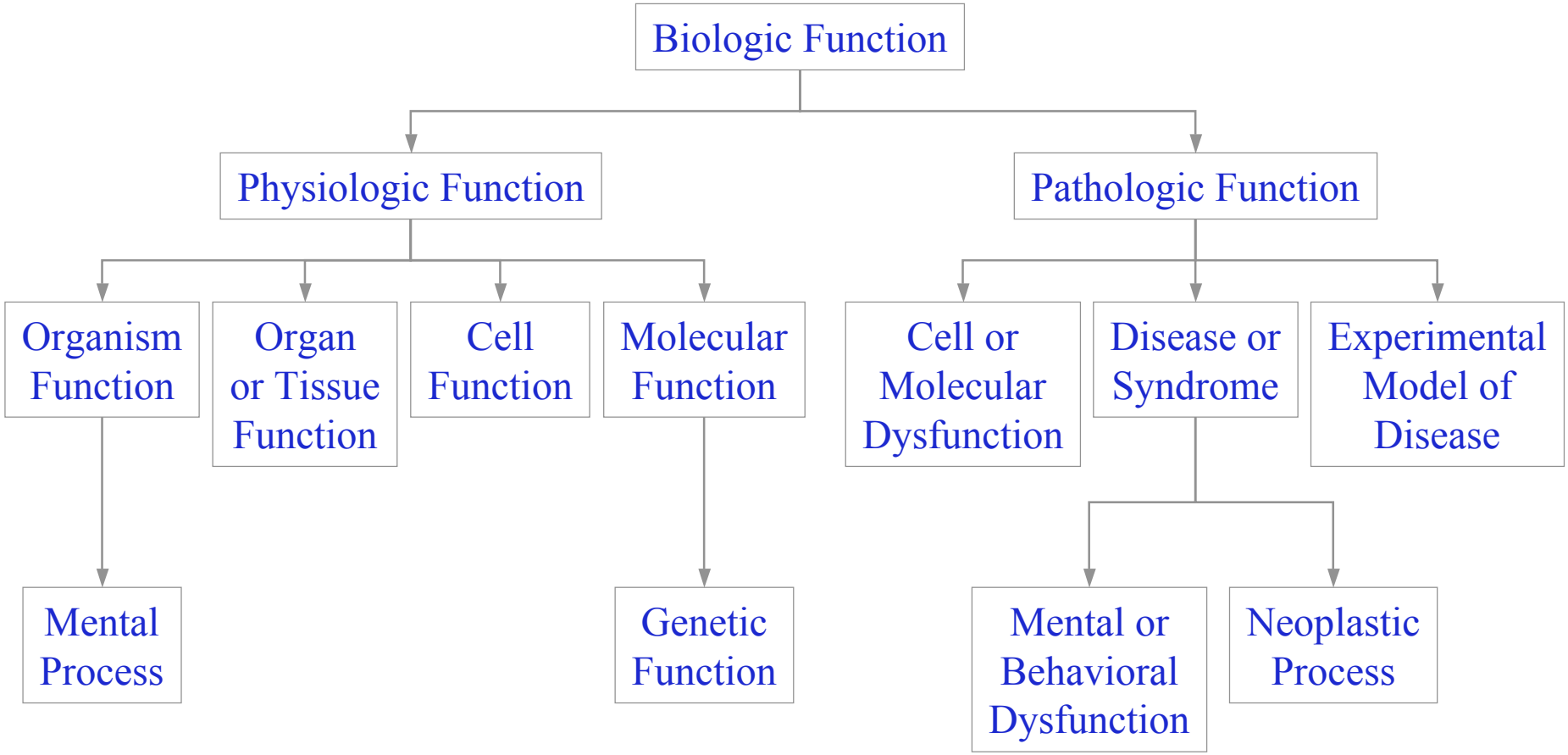
“Biologic Function” hierarchy (isa)



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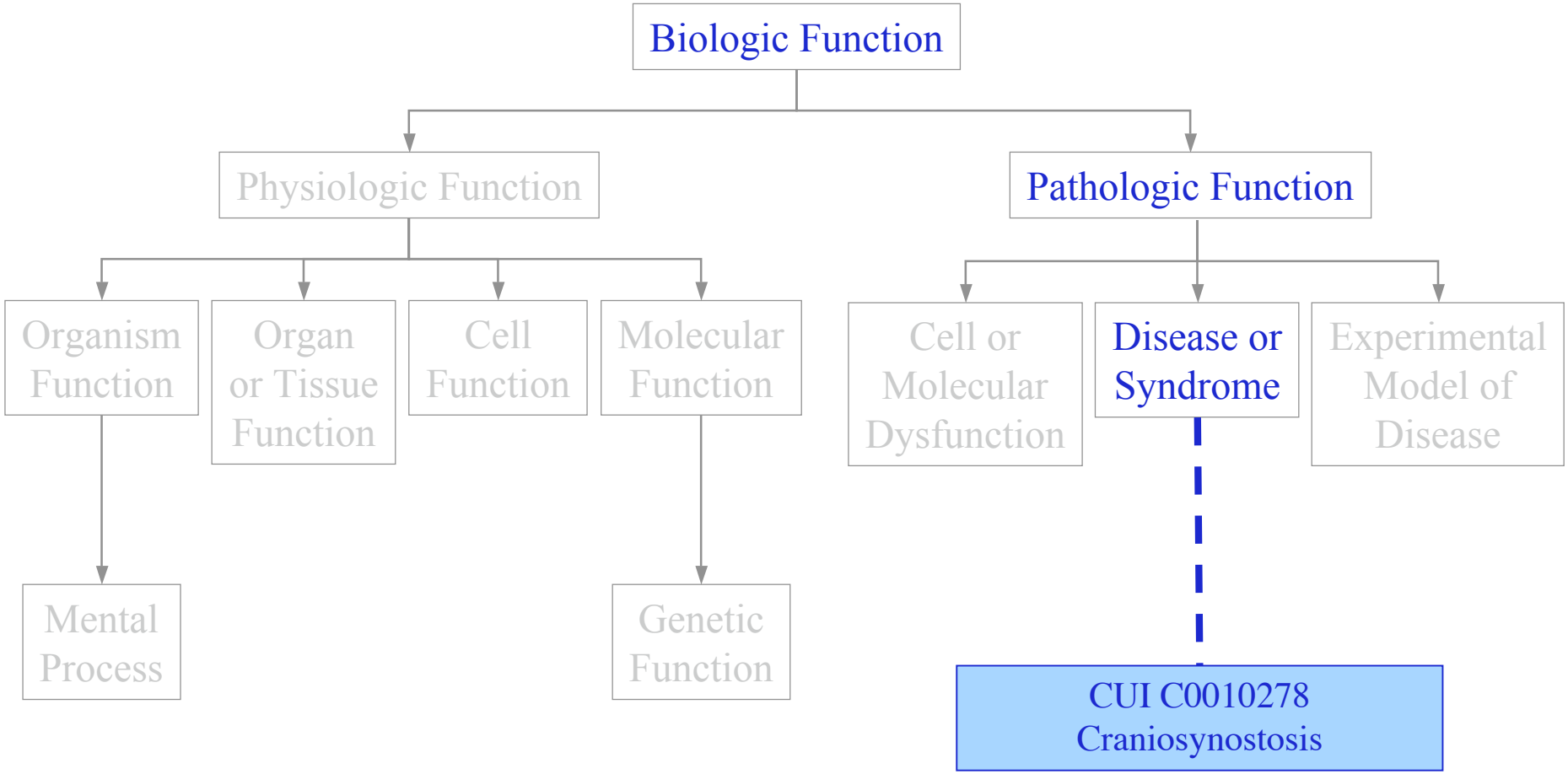
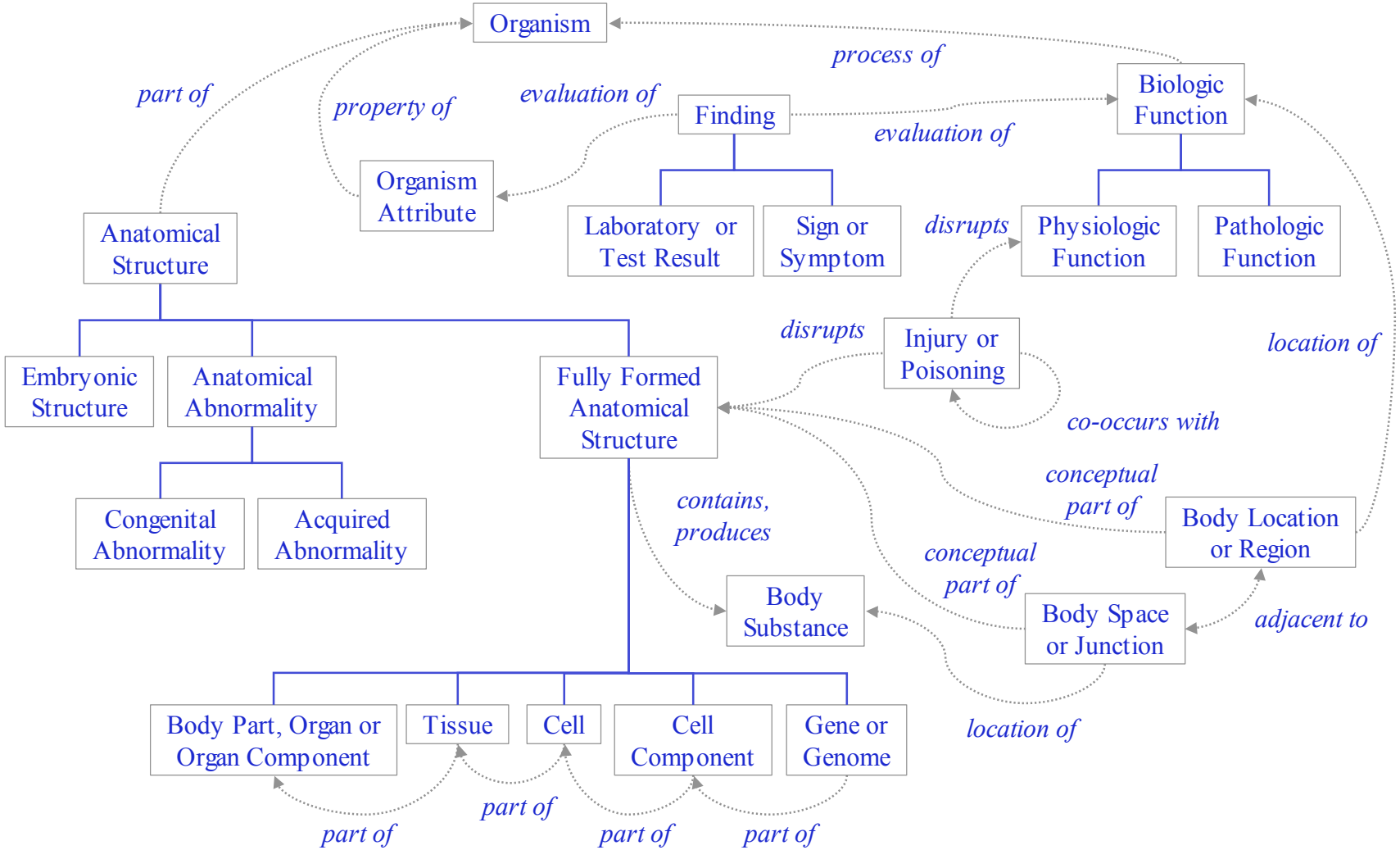


Overview of the Semantic Network



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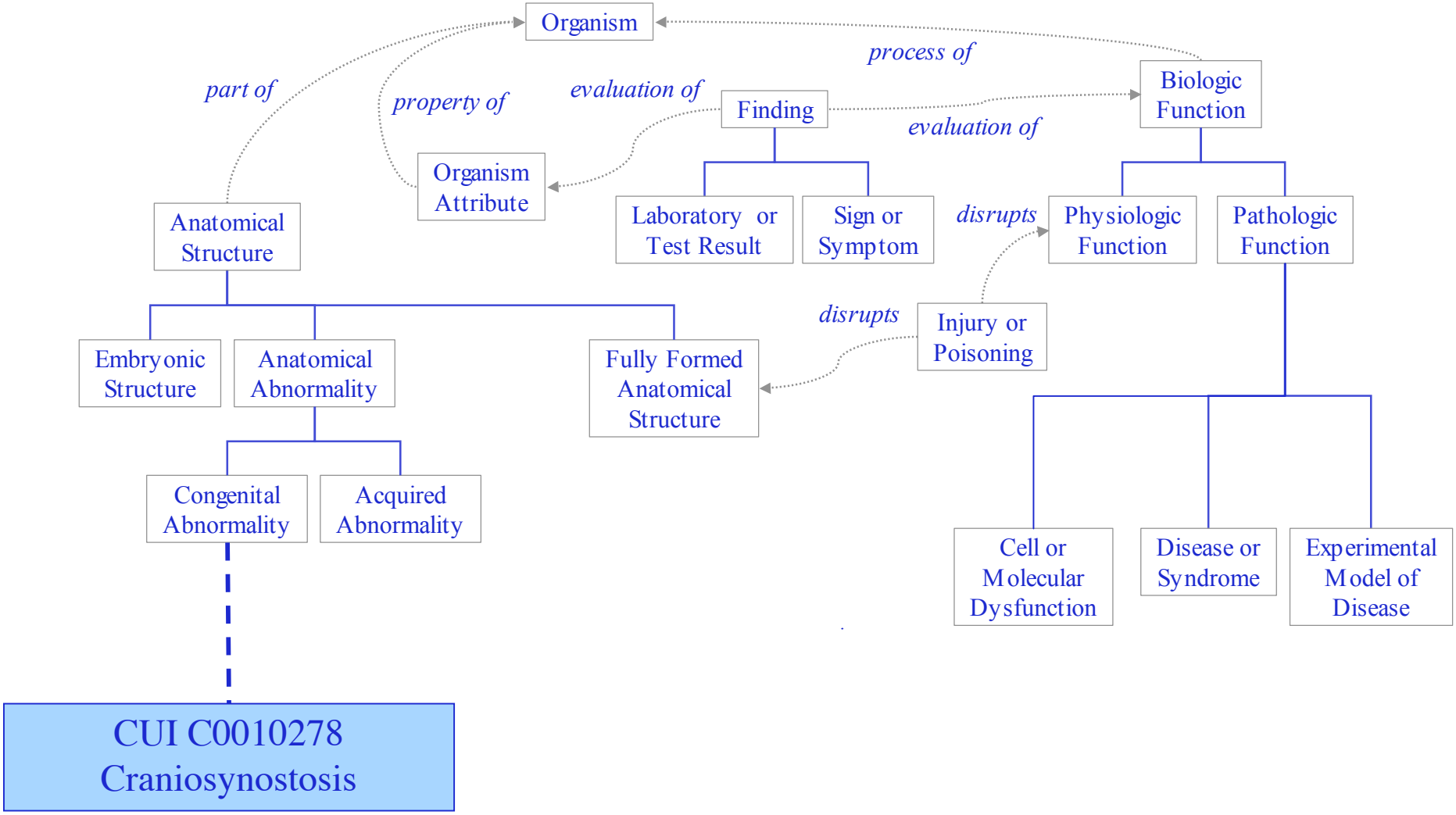
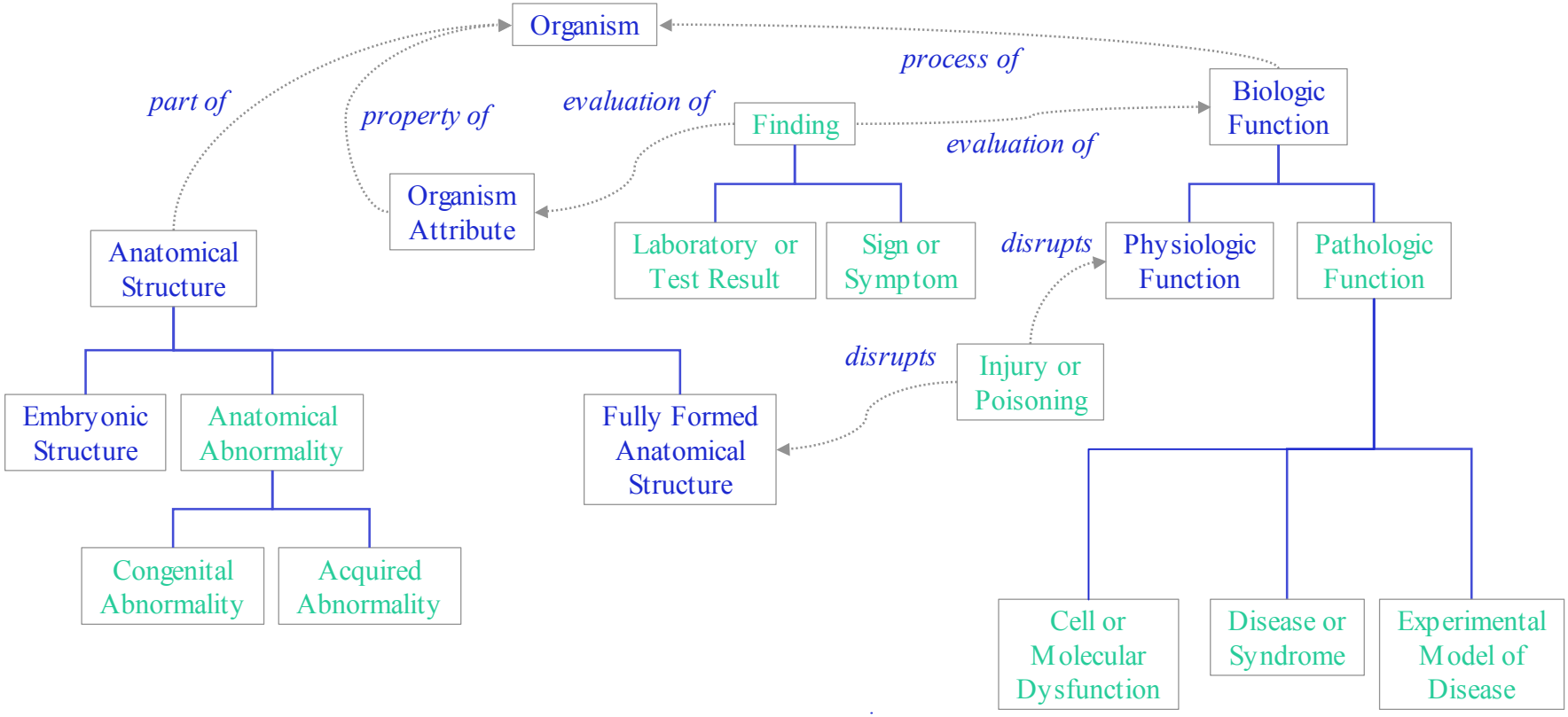
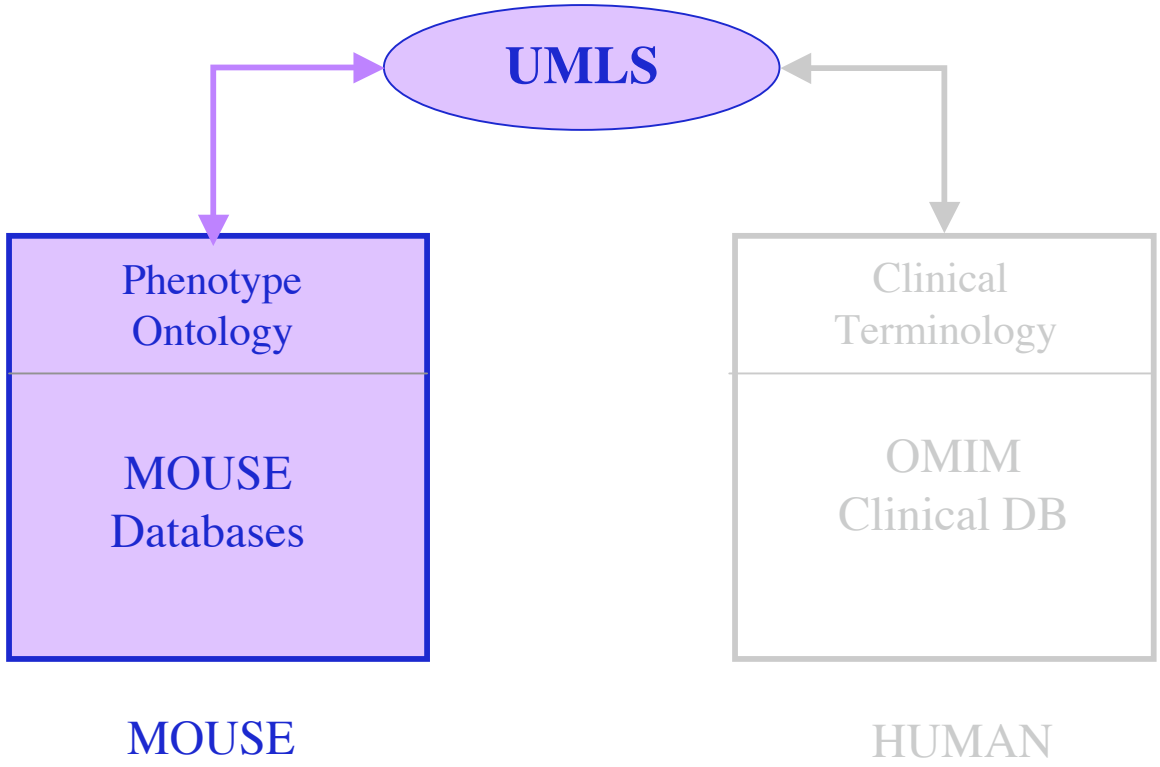


Semantic Groups



Disorders

Phenotype coding: role of the UMLS



Mammalian Phenotype Ontology (MPO)

- 14,662 terms
- 6,307 concepts
- MP:0003561 Rheumatoid arthritis
- MP:0000218 increased leukocyte cell number
  increased leukocyte count
  increased WBC count
  increased WBC number
  increased white blood cell number
  leukocytosis
- MP:0000410 waved hair
  curly hair
  waved fur
  wavy hair

Mapping to UMLS (1/3)

- Step 1 : Exact/normalized match
- Results
  - 2,065 MPO terms mapped successfully(14%)
  - 1,495 MPO concepts mapped successfully (24%))
  - Among them, 1,432 correspond to Disorders in UMLS (SG)
- Examples mapped successfully
  - MP:0000062 increased bone density-> C1141880 Bone density increased (NSI)
  - MP:0000081 craniostosis -> C0010278 Craniosynostosis (syn in MPO, EM)
  - MP:0000061 brittle bones -> C0029434 Osteogenesis Imperfecta (syn in UMLS, EM)
- Examples unmapped
  - MP:0000100 abnormal ethmoidal bone
  - MP:0000101 absent ethmoidal bone
  - MP:0000687 small lymphoid organs
  - MP:0000689 abnormal spleen structure

Mapping to UMLS (2/3)

- 11,466 unmapped terms (4,812 concepts)
  - MP:0000100 abnormal ethmoidal bone
  - MP:0000101 absent ethmoidal bone
  - MP:0000687 small lymphoid organs
  - MP:0000689 abnormal spleen structure
- Step 2 : demodification
  - 30 modifiers: abnormal, absent, small…..
  - Demodified terms
  - Mapping to UMLS
- Results : demodified terms
  - 9,845 <<modifier> xxx> terms in MPO
  - 7,925 unique terms (after demodification)

Mapping to UMLS (3/3)

- Results : mapping after demodification
  - 2,359 MPO terms mapped successufully after demodification (out of 11,466 , 20%)
  - Unique terms : 1,586 (20%)
  - 1,645 MPO concepts mapped successufully after demodification (out of 4,812, 34%)
- Demodified terms correspond mostly to:
  - Anatomical concepts
    - Semantic Group Anatomy in UMLS
    - 1410 terms, e.g., abnormal <anatomical structure>
    - MP:0000005 (increased) brown fat -> C0006298 Brown Fat (Tissue, ANAT)
  - Physiology
    - Semantic Group Physiology in UMLS
    - 516 terms, e.g., abnormal <physiological process>
    - MP:0000057 (abnormal) osteogenesis -> C0029433 Osteogenesis (Organ or Tissue Function, PHYS)

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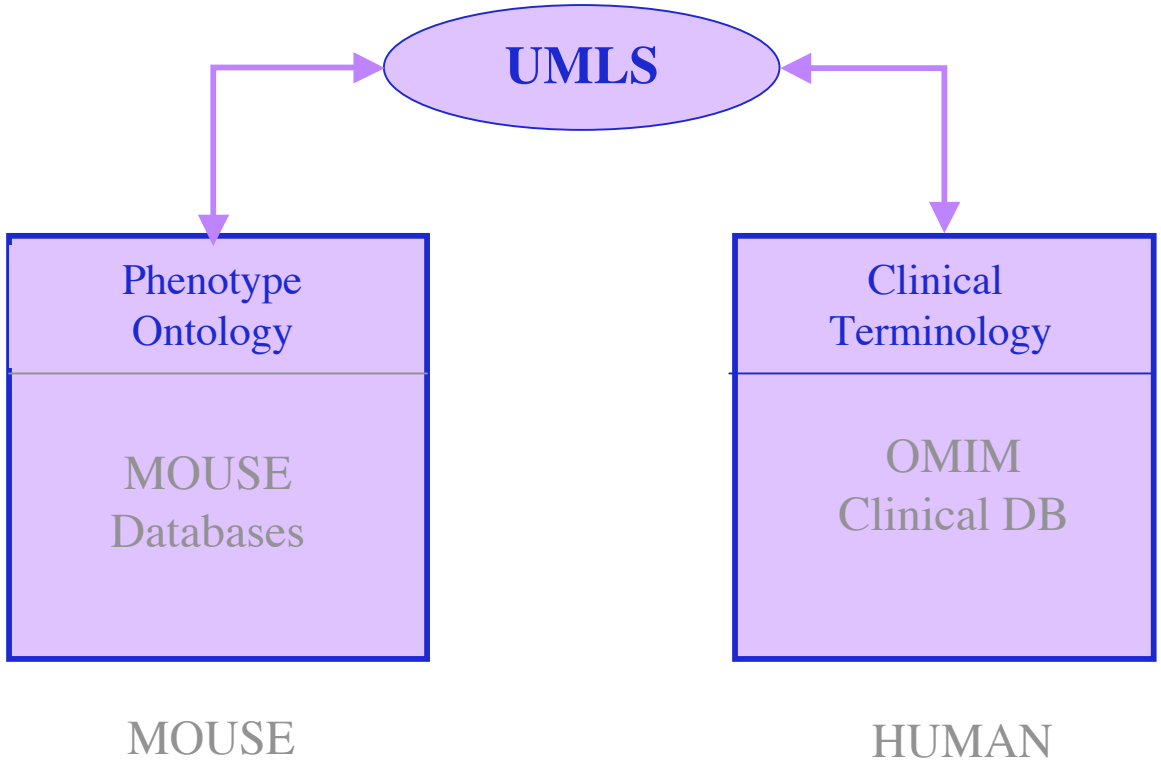


Discussion

- Compositionality in phenotype terms

< abnormal <anatomical structure > >

30 modifiers

Functional Concept

Qualitative Concept

abnormal
decreased
increased
reduced
small …..

Anatomy

Physiology

SGs

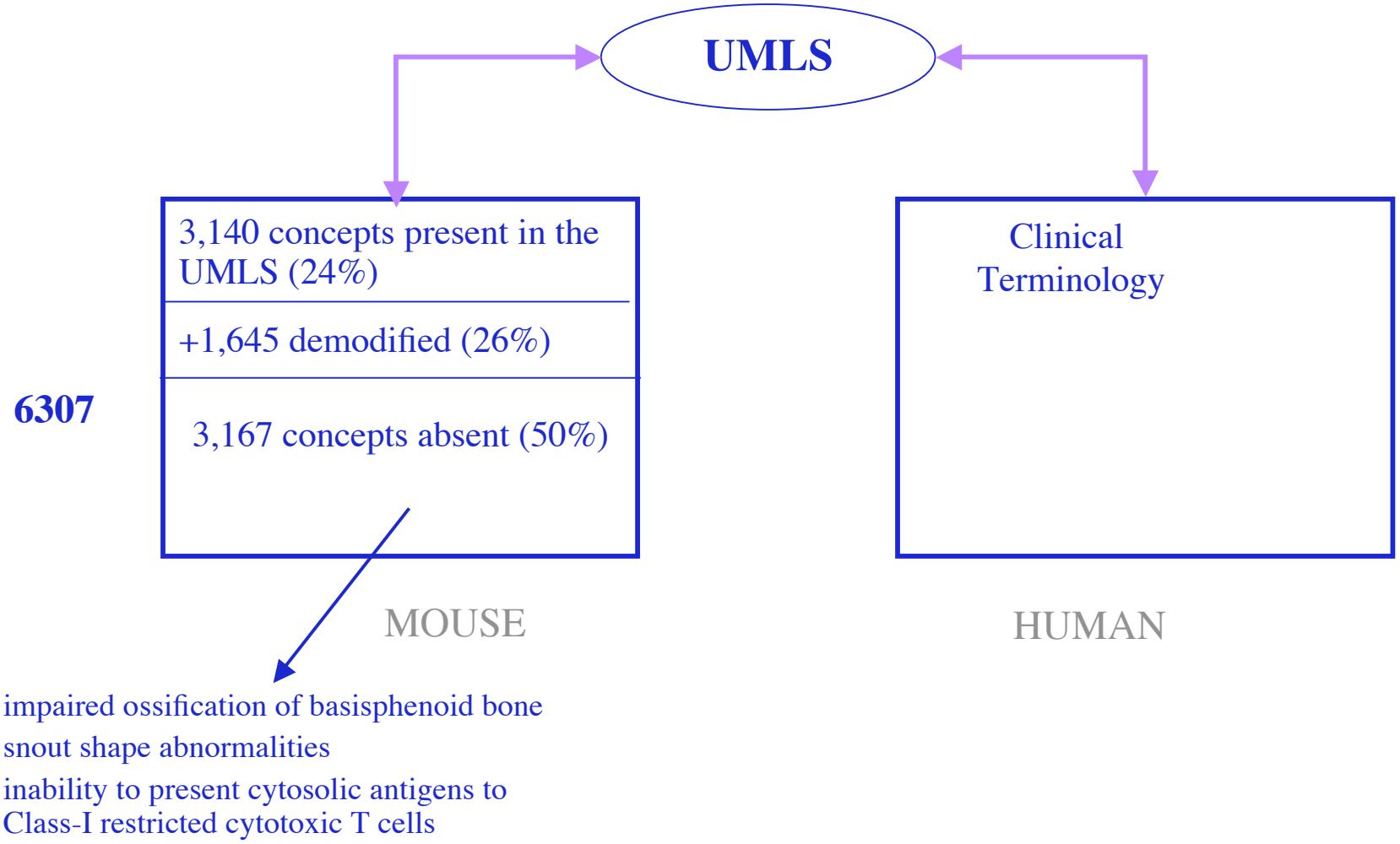
Class Name: abnormal (**PATO:0000460**)
Is A: deviation(from_normal)
Synonym: aberrant, atypical, defective
[R]Is A: pathological

entities from any ontology

Phenote : EQ model : combine entities from any ontology
with qualities / traits (such as those in PATO)

Discussion

- Role of the UMLS in integrating phenotype terminologies



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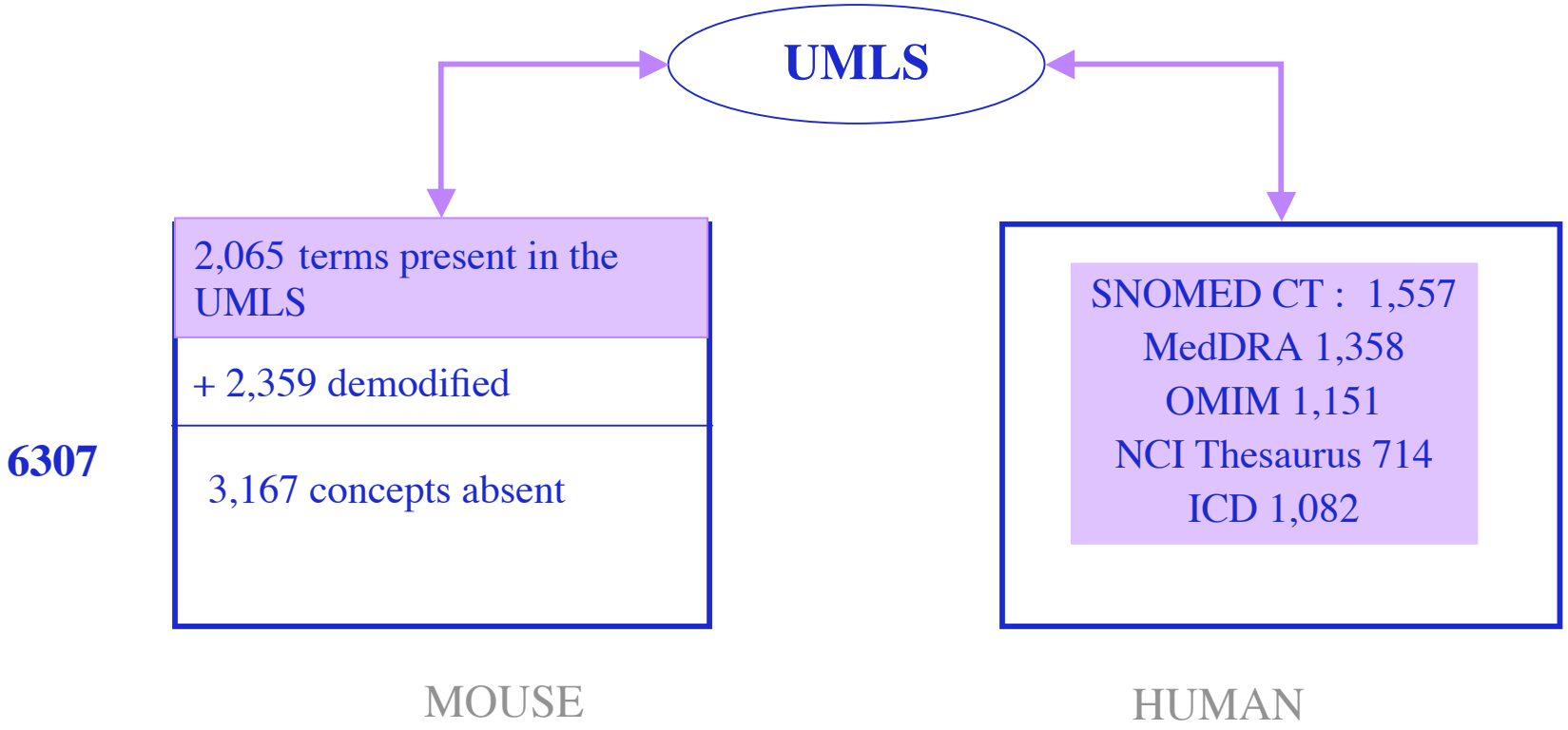
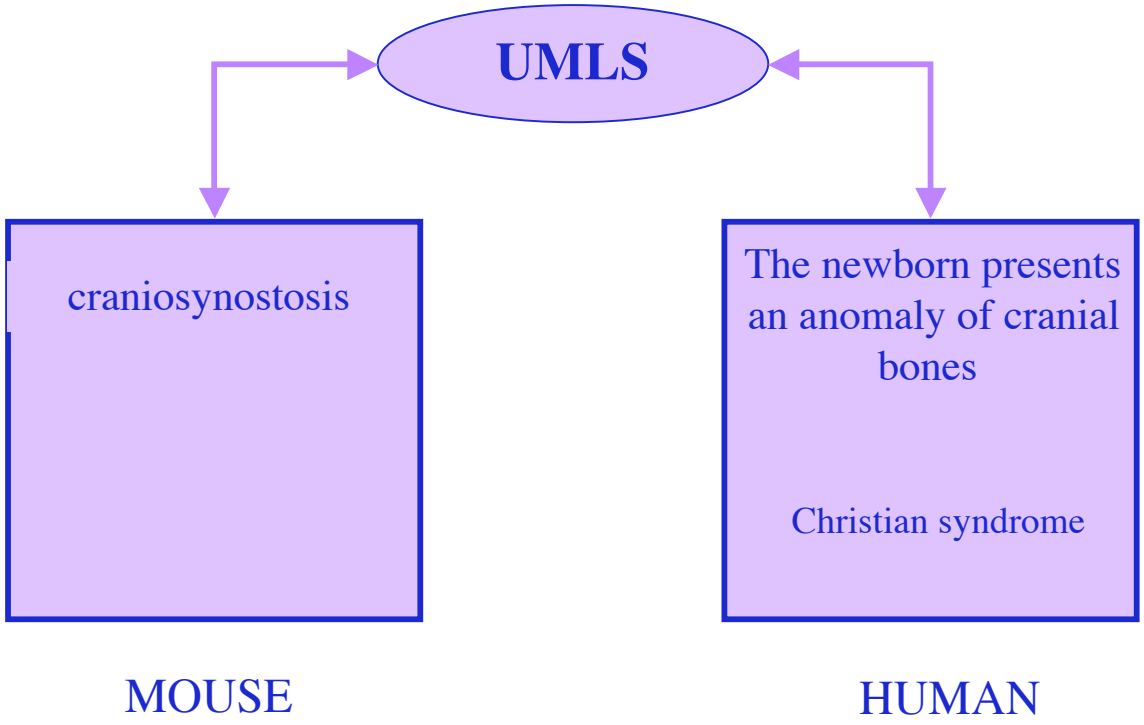


Discussion

- Role of the UMLS in integrating phenotype data
  - OMIM, Clinical DBs, Mouse DBs



Acknowledgements

- Olivier Bodenreider, NLM
- Fleur Mougin, ISPED

- Download/ customize/ browse the UMLS
- Knowledge Source Server

- **umls**ks.nlm.nih.gov/